



SLC20A2 gene

solute carrier family 20 member 2

Normal Function

The *SLC20A2* gene provides instructions for making a protein called sodium-dependent phosphate transporter 2 (PiT-2). This protein plays a major role in regulating phosphate levels within the body (phosphate homeostasis) by transporting phosphate across cell membranes. Phosphate is needed for many functions including the breakdown of substances (metabolic processes), signaling between cells, and the production of DNA building blocks (nucleic acids) and fats. PiT-2 uses positively charged sodium atoms (ions) to transport phosphate across the cell membrane.

Health Conditions Related to Genetic Changes

familial idiopathic basal ganglia calcification

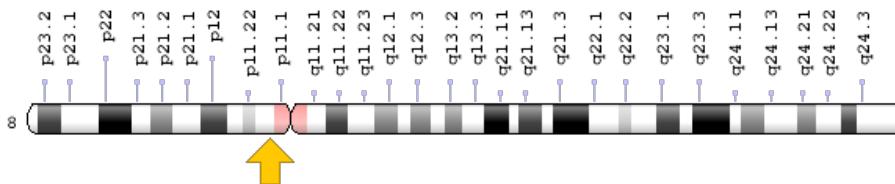
Approximately 20 *SLC20A2* gene mutations have been found to cause familial idiopathic basal ganglia calcification (FIBGC). This condition is characterized by abnormal deposits of calcium (calcification) in the brain and movement and psychiatric problems. Most of the mutations that cause FIBGC change single protein building blocks (amino acids) in the PiT-2 protein and severely impair its ability to transport phosphate into cells. As a result, phosphate levels in the bloodstream rise. In the brain, the excess phosphate combines with calcium and forms deposits.

Although the *SLC20A2* gene is active (expressed) throughout the body, its activity is highest in the basal ganglia and other brain regions that are involved in FIBGC, which may explain why the effects of these mutations are limited to these regions.

Chromosomal Location

Cytogenetic Location: 8p11.21, which is the short (p) arm of chromosome 8 at position 11.21

Molecular Location: base pairs 42,416,462 to 42,542,213 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- gibbon ape leukemia virus receptor 2
- GLVR-2
- GLVR2
- MLVAR
- murine leukemia virus, amphotropic, receptor for
- PIT-2
- PIT2
- S20A2_HUMAN
- sodium-dependent phosphate transporter 2
- solute carrier family 20 (phosphate transporter), member 2

Additional Information & Resources

GeneReviews

- Primary Familial Brain Calcification
<https://www.ncbi.nlm.nih.gov/books/NBK1421>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC20A2%5BTIAB%5D%29+OR+%28%28PiT-2%5BTIAB%5D%29+OR+%28PiT2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER FAMILY 20 (PHOSPHATE TRANSPORTER), MEMBER 2
<http://omim.org/entry/158378>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC20A2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC20A2%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10947
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6575>
- UniProt
<http://www.uniprot.org/uniprot/Q08357>

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